



estonian genome project



**CARTaGENE  
Project**



**CIGMR**

# Public Population Project in Genomics (P<sup>3</sup>G)

**Send correspondence to:**

Bartha Maria Knoppers

**Chair**

Centre de recherche en Droit Public

Université de Montréal

3101, Chemin de la tour

Montréal, (QC) H3T 1J7

**knoppers@droit.umontreal.ca**

**Phone:** +1 (514) 343-6714

**Fax:** +1 (514) 343-2122

Mylène Deschesnes, *Executive Director*

Isabel Fortier, *Knowledge database Director*

Thomas J. Hudson

**Chief Scientific Officer**

McGill University and

Genome Quebec Innovation Centre

740 Dr. Penfield Avenue, Room 7105

Montréal, (QC), H3A 1A4

**tom.hudson@mcgill.ca**

**Phone:** +1 (514) 398-3311 ext. 00385

**Fax:** +1 (514) 398-2262

<http://www.p3gconsortium.org>



# Overview

**The *Public Population Project in Genomics* (P<sup>3</sup>G) is a not-for-profit and non commercial organization building an international consortium to foster collaboration between researchers in the field of population genomics.**

## **Our Vision:**

Foster international harmonization for public projects in population genomics.

## **Our Goals:**

- Understand the interaction between genes, environment, lifestyle and disease
- Create the engine for the transfer of this knowledge to health care systems

## **Our Approach:**

International collaboration so as to ensure public access to population genomics data according to prevailing ethical and legal norms.



# Overall Objectives of P<sup>3</sup>G

## Objectives

**Provide necessary coordination and harmonization for data collection, production, and storage, to facilitate international collaboration, advance science and maximize public health benefits**

**Develop common understanding of the socio-ethical and legal issues**

**Build and maintain a knowledge community for information sharing through a database infrastructure**

**Foster a deeper understanding of the relative contribution of genetic and non-genetic determinants to health and disease**

**Transfer this knowledge to the international community so as to optimize benefits for public health care worldwide**



## Deliverables

**Common nomenclature and communication tools required for seamless data sharing**

**Ethical and legal framework for population-based projects**

**Public data in a P3G knowledgebase leading to exchange of scientific information and new scientific discoveries in population health research**

**Discovering linkages between genetics and health**

**Global access to common data including by developing countries**



## Partner Projects

### GenomEUtwin

**Study 800,000 twin pairs from a collaborative European pool of registries through a combination of genetic, epidemiology and phenotype data for common diseases**

### Estonian Genome Project

**Collect data from up to one million participants into a database, including health status, genomic DNA, plasma and genealogical data**

### CARTaGENE

**Obtain personal, medical and sociological data and biological samples from 50,000 random participants for the study of normal genetic variation**

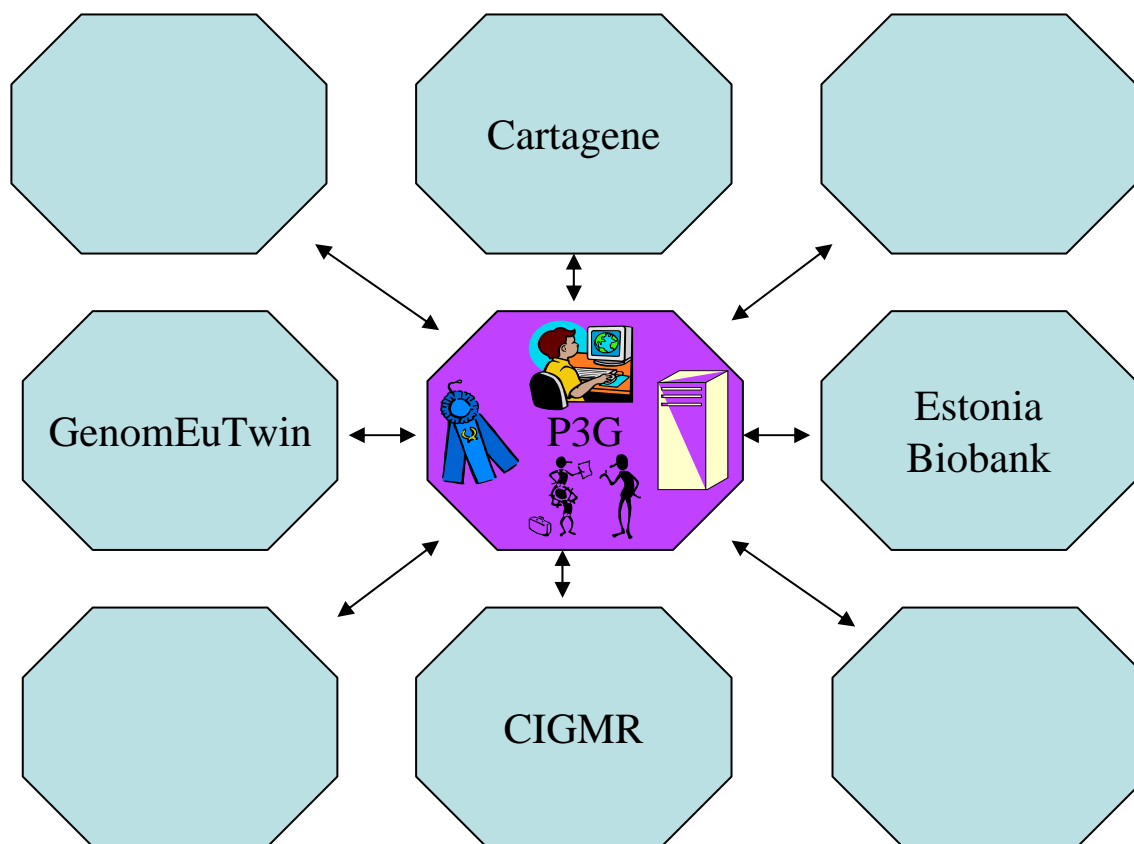
### CIGMR

**Houses several disease specific population and family based cohorts, and has longitudinal data on over 20,000 individuals**



# Consortium Model

An international resource for the coordination and exchange of ideas and data that will be generated by the various population biobanks





# Areas of Mutual Interest

- **Harmonization** of biological, medical, demographic and social data collected from participants so as to use this information synergistically **in developing effective health care solutions**
- Coordination of possible **access between population databanks** while protecting confidentiality subject to legal constraints, ethical review and governance
- Development of **security measures** for the protection of genetic data and banks in accordance with the highest international standards
- Sharing of approaches to **ethics, public engagement, governance and intellectual property issues**
- **Exchange of experts and young researchers** in many human and social scientific disciplines in addition to biology, public health and genetics
- Determining common biological phenotypes as well as **agreement on methods and quality control**
- Developing **common nomenclature** for data obtained in population genomics projects
- Coordinated development of **bioinformatics** for compatible data mining and for clarity on ownership/copyright issues
- **Support for the transfer of knowledge and technology** to other developed or developing countries
- **Comparative evaluation and validation of research results** and/or hypotheses on health and disease
- **International leadership** in the ethics of genomic and genetic research involving populations



# Helsinki/Tallinn Workshops

Goal: Establishing the Work Plan for 2004-2006

## **I. Socio-Demographic Core**

Claude Laberge  
Dorret Boomsma

Canada  
Netherlands

## **II. Phenotype Core**

Francois Cambien

France

## **III. Storage, Genotyping and Logistics Core**

Leena Peltonen  
Andres Metspalu

Finland  
Estonia

## **IV. Public Engagement, Ethics and Governance Core**

Anne Cambon-Thomsen  
Jennifer Harris  
Beatrice Godard  
Ruth Chadwick

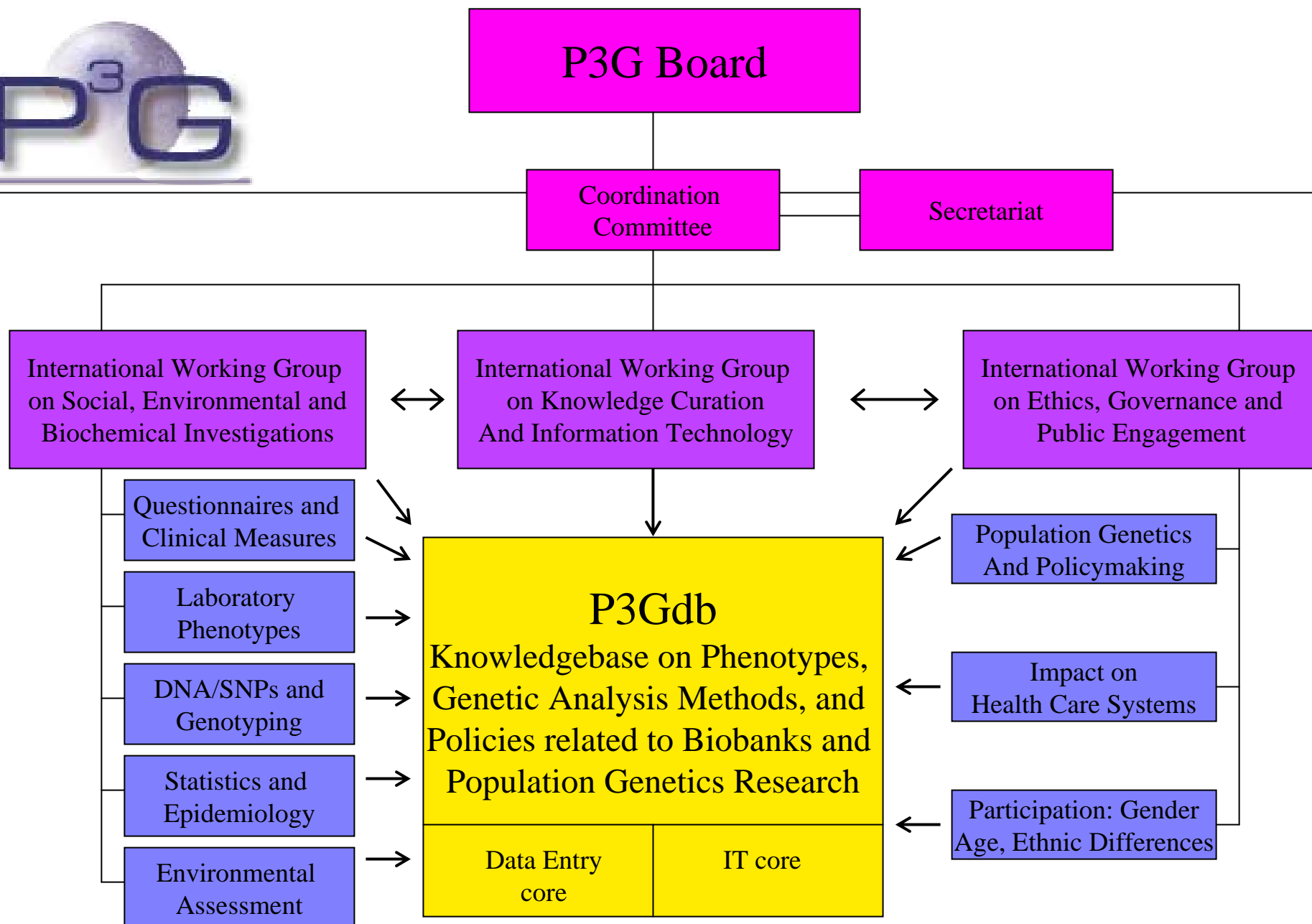
France  
Norway  
Canada  
United Kingdom



## Major Outcomes of the Helsinki/Tallinn/ASHG

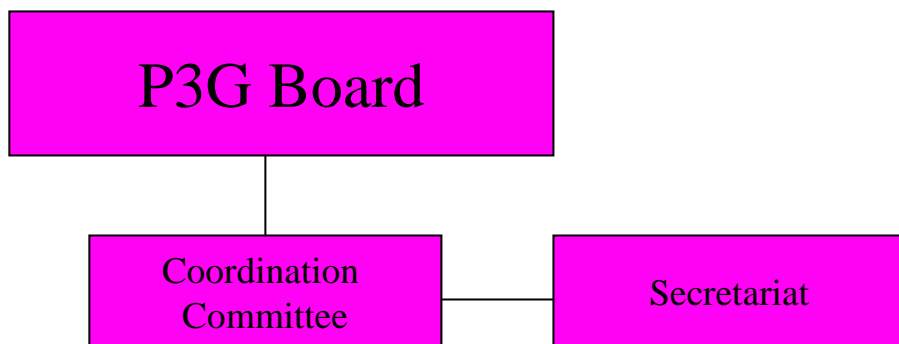
- Conclusion that the largest need at present is in better refining phenotypes as opposed to genotypes
- Recommendation to invest early in the creation of a knowledgebase that will track large-scale population genetic studies (including biobanks) - and record their characteristics in order to benefit from existing knowledge and compare approaches (biomedical, ethical, etc)
- Recommendation to merge advisory committees (for easier coordination)
- Definition of core activities for 2004-2006







# P3G Governance

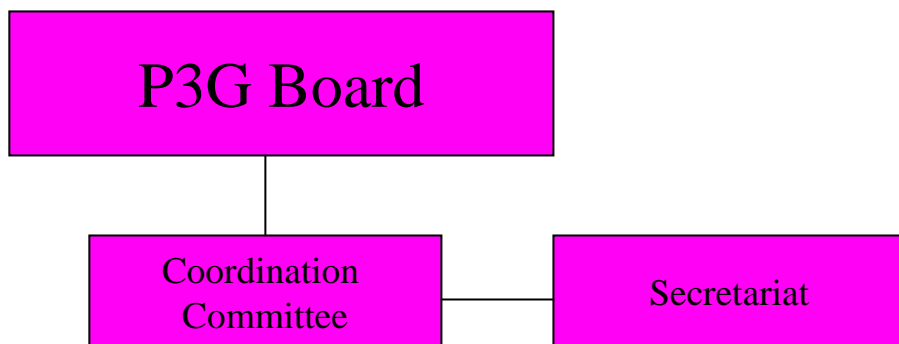


## Role of Board :

- ❖ Assure that P3G activities meet the stated goals of harmonization, communication, and public access of P3G deliverables
- ❖ Communication among partners (and funding agencies)
- ❖ Accountability of P3G fund



# P3G Governance

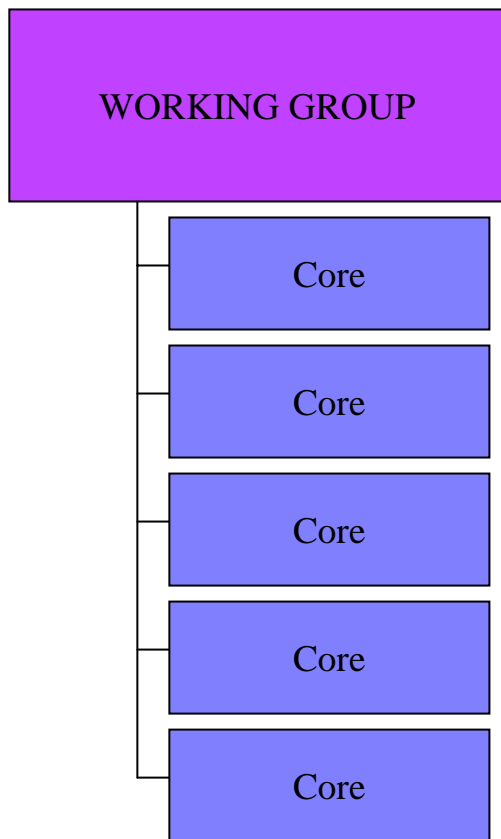


## **Coordination Committee :**

- ❖ Composition includes leaders of Working Group and Cores and BioBank representatives
- ❖ Key role in managing P3G activities, assuring coordination, addressing new issues, etc.
- ❖ Organization of P3G symposia, workshops, exchanges
- ❖ Creating a P3G training program
- ❖ Supported by a small administrative staff



# P3G Working Groups and Cores



## Working Group

- ❖ International and Multidisciplinary Team
- ❖ Assures coordination among cores via quarterly discussions, annual meeting
- ❖ Sets priorities for cores

## Cores

- ❖ Principal work units of P3G, focused on specific issues related to biobanks.
- ❖ In most (but not all) cases, cores are hosted at a single site and are integrated with an existing team actively involved with a biobank.



# International Working Group on Social, Environmental and Biochemical Investigations

International Working Group  
on Social, Environmental and  
Biochemical Investigations

Questionnaires and  
Clinical Measures

Laboratory  
Phenotypes

DNA/SNPs and  
Genotyping

Statistics and  
Epidemiology

Environmental  
Assessment

- ❖ Early Focus on Risk Factors for Cardiovascular Phenotypes in order to capitalize on existing expertise, yet face new opportunities made available by new technologies in phenotyping, environmental measures, genomics, etc.
- ❖ 6 month goals - collate information related to existing studies, available phenotype information, etc., and deposit in P3Gdb
- ❖ Interim Leader: Erich Wichmann (Germany)



# Selected notes in regards to the Social/ Environmental/Biochemical Working Group

Questionnaires and  
Clinical Measures

- ❖ Collation of information related to existing studies
- ❖ Evaluation of socio-demographic information, collected for public health purposes
- ❖ Definition of clinical phenotypes with different degree of complexity

Laboratory  
Phenotypes

- ❖ Short term Focus: Storage of plasma, serum and urine for downstream studies
- ❖ Research Focus on intermediate phenotypes, including new methodologies in proteomics and metabolomics

DNA/SNPs and  
Genotyping

- ❖ Quality assessment measures related to DNA and genotyping

Statistics and  
Epidemiology

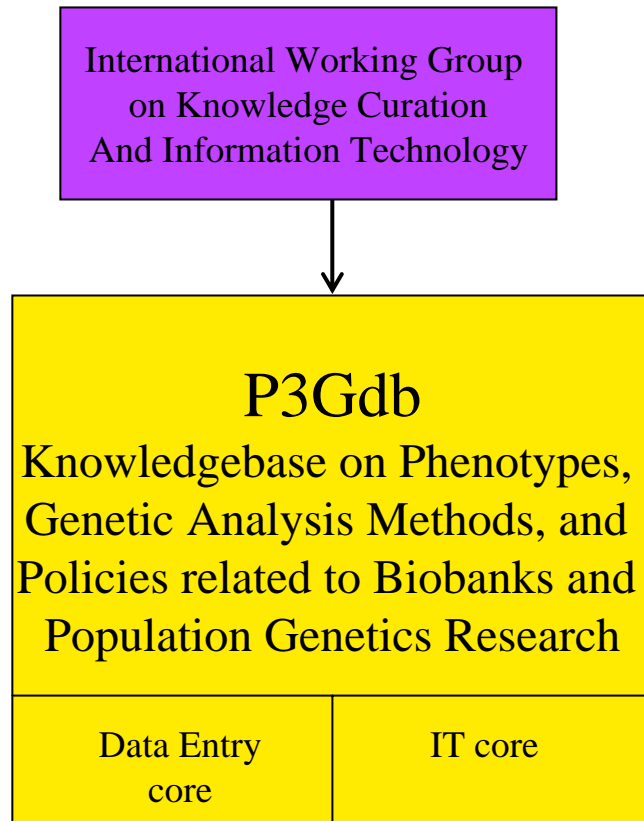
- ❖ Tracking of risk alleles and haplotypes
- ❖ Research into methods that allow merging and comparing population genetics studies that offer more power than “reducing” the data to a small set of “common” phenotypes

Environmental  
Assessment

- ❖ Evaluation of environmental parameters needed for population genetics
- ❖ Definition of a core set of environmental information for studies of gene-environment interaction



# International Working Group on Knowledge Curation And Information Technology



- ❖ P3G db is needed, as there is no existing resource that captures and disseminates information on large-scale epidemiological studies. Most of the details regarding questionnaires, phenotypes, methods are not available electronically (or at all), making comparative analyses difficult, and hindering meta-analyses (that could increase power, generate new hypotheses, etc.).
- ❖ “OMIM”-type resource.
- ❖ All activities of P3G, from all cores, must be organized and disseminated widely
- ❖ Multidisciplinary expertise required in informatics and communication.
- ❖ Data entry will be prioritized by working groups and cores.
- ❖ Immediate scientific benefits: Increased interactions between and beyond P3G as soon as P3Gdb is operational
- ❖ Interim Leader: Jan-Eric Litton (Sweden)



# International Working Group on Ethics, Governance and Public Engagement

International Working Group  
on Ethics, Governance and  
Public Engagement

Population Genetics  
And Policymaking

Impact on  
Health Care Systems

Participation: Gender  
Age, Ethnic Differences

- ❖ Public Engagement, Ethics, Public welfare (prevention/promotion) need to be integrated
- ❖ Interim Leader: Béatrice Godard (Canada)
- ❖ Policy issues not only related to the development of biobanks, but the translation of new data for the benefit of the public
- ❖ What risks and benefits will arise due to current practices in commercializing genetic information
- ❖ Research into practices used for public engagement
- ❖ Better understanding of public perception as they relate to socio-cultural issues





# Challenges and Opportunities in Funding P3G

## **Challenges:**

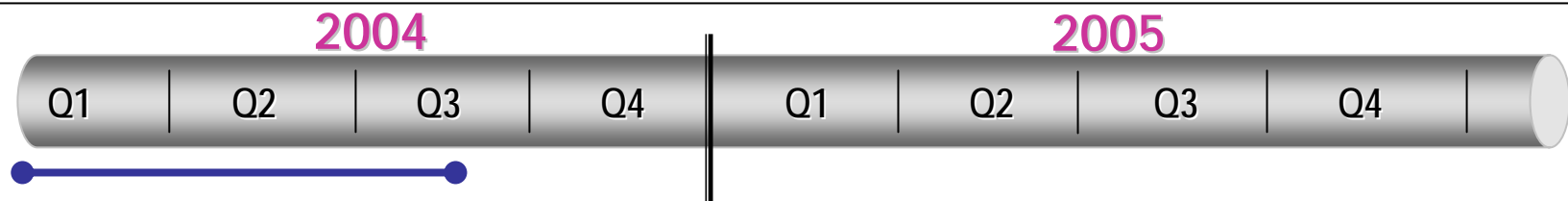
- ❖ Multi-national research initiatives are difficult due to limitations in funding research beyond national/continental boundaries.
- ❖ Biobanks are expensive - additional costs for international coordination are difficult to attract when biobank groups are working at raising funds to initiate or maintain a national Biobank.

## **Opportunities:**

- ❖ Scientific benefits: Increased power afforded by large-scale collaboration, larger cohort-sizes for gene-environment studies, rarer phenotypes or subsets of participants with value-added phenotypes.
- ❖ Cost efficiencies: New knowledge (such as lower cost laboratory phenotyping methods) can decrease costs of biobank projects.
- ❖ Funding agencies may be more willing to fund portions of P3G activities (such as a core, for example) in the context of a larger effort with international co-funding).



# Timeline (High Level)



## Phase 1 – Planning and Funding

- Memorandum of Understanding completed
- Development of membership rules is complete
- Communication globally with participating projects and enrolment of new members are ongoing
- Writing of a detailed project plan for funding is in progress
- Securing of funding for global operations is ongoing
- Work plan for 2004 to 2006 has been drafted

## Phase 2 – Implementation

- Creation of Board, Secretariat and Coordinating Committee
- Creation of detailed implementation plan for knowledge base
- Creation of forecast/deliverables tracking system
- Continue to enroll new members

## Phase 3 – Deliverables

- Strengthen communications
- Continue to enroll new members
- Collection of common data
- Promotion, marketing, communications



## **Next Steps:**

- Build Knowledge Database
- Membership drive
- Identify Remaining Cores
- Funding (Major) Applications
- Canadian Cohorts Meeting (May, 2005)
- IWG: Ethics, Governance, Public Engagement Workshop (Sept. 2005)
- Board of Directors Meeting (U.K., Sept. 2005)